OMIM Home | Search | Comments

*151460 PROTEIN-TYROSINE PHOSPHATASE, RECEPTOR-TYPE, C; PTPRC

Alternative titles; symbols

LEUKOCYTE-COMMON ANTIGEN; LCA
T200 GLYCOPROTEIN; CD45
Ly5, HUMAN HOMOLOG OF
B220
SEVERE COMBINED IMMUNODEFICIENCY DUE TO PTPRC DEFICIENCY, INCLUDED
SCID DUE TO PTPRC DEFICIENCY, INCLUDED

TABLE OF CONTENTS

Database Links

MEDLINE | Protein | DNA | LocusLink | Gene Map | GDB | Nomenclature

Gene Map Locus: 1q31-q32

Note: pressing the symbol will find the citations in MEDLINE whose text most closely matches the text of the preceding OMIM paragraph, using the Entrez MEDLINE neighboring function.

TEXT

T200 glycoprotein, also known as leukocyte-common antigen (LCA), or CD45, is a major high molecular weight leukocyte cell surface molecule. It is an integral membrane protein tyrosine phosphatase (Charbonneau et al., 1988; Tonks et al., 1988, 1990). It is expressed on all hematopoietic cells except mature red cells and their immediate progenitors. It is not found, however, on other differentiated tissues; thus, it can be used as an antigenic marker with which to identify undifferentiated hematopoietic tumors. Ralph et al. (1987) isolated cDNA clones of T200 glycoprotein from a variety of human lymphoid cells, deduced the complete primary structure of the molecule from the cDNA sequence of these clones, and identified 3 structural variants which probably arise by cell-type-specific alternative splicing.

Trowbridge (1991) reviewed the information on CD45 indicating that it is a prototype for transmembrane protein-tyrosine phosphatase (PTP). Fischer et al. (1991) reviewed protein-tyrosine phosphatases in general and CD45 specifically. CD45 is found only in hematopoietic cells where it comprises up to 10% of the cell surface. It is a prime example of a receptor-linked PTP of type I.

Irie-Sasaki et al. (2001) showed that CD45 suppresses JAK kinases (see 147795) and negatively regulates cytokine receptor signaling. Targeted disruption of the CD45 gene leads to enhanced cytokine and interferon receptor-mediated activation of JAKs and STAT proteins. In vitro, CD45 directly dephosphorylates and binds to JAKs. Functionally, CD45 negatively regulates interleukin-3-mediated cellular proliferation, erythropoietin-dependent hematopoiesis, and antiviral responses in vitro and in vivo. Irie-Sasaki et al. (2001) concluded that their data identified an unexpected and novel function for CD45 as a hematopoietic JAK phosphatase that negatively regulates cytokine receptor signaling.

By in situ hybridization, Ralph et al. (1987) demonstrated that the gene encoding human T200 is located on chromosome 1q31-q32. By somatic cell hybridization, Akao et al. (1987) confirmed the chromosomal assignment. Fernandez-Luna et al. (1991) isolated the CD45 gene in a single YAC clone and estimated its size to be approximately 120 +/- 10 kb. By physical mapping on a 610-kb YAC, Goff et al. (1999) determined that the PTPRC gene colocalizes with marker D1S413 on chromosome 1q31-q32.

The smallest human T200 variant is homologous to Ly5 in the mouse (Saga et al., 1986). Chromosome 1 of the mouse was found to be the site of the gene or genes for at least 2 isoforms of Ly5 (Shen et al., 1985). Seldin et al. (1987) studied variants of Ly5 in inbred and natural populations of mice. Summarizing the data, Seldin et al. (1987) stated that 'genetic and biochemical data favor the interpretation that a single gene on distal chromosome 1 (of the mouse) encodes these Ly-5 isoforms.' The gene is located in a region of the distal part of mouse chromosome 1 that carries many genes homologous to genes in human 1q21.3-q32 (Seldin et al., 1988). Seldin et al. (1988) stated that CD45, the human equivalent of Ly5, is located in band 1q31. They commented on the large number of genes of immunologic interest clustered in this region. The list includes the Ly17 gene, which encodes the Fc IgG1/IgG2A receptor (Ravetch et al., 1986); the IGFR2 gene (146790) has been mapped to human chromosome 1. Thomas et al. (1987) presented evidence that variants of T200 glycoprotein are generated in the mouse by alternative mRNA splicing.

The CD45 gene contains 35 exons. The receptor is essential for the activation of T and B cells by mediating cell-to-cell contacts and regulating protein-tyrosine kinases involved in signal transduction. CD45 is also involved in integrin-mediated adhesion and migration of immune cells. The glycoprotein exists in multiple isoforms, depending on alternative splicing of exons 4, 5, and 6. The corresponding protein domains are characterized by the binding of monoclonal antibodies specific for CD45RA (exon 4), CD45RB (exon 5), CD45RC (exon 6), and CD45RO (exons 4 to 6 spliced out). In T cells, the alternative splicing of CD45 is regulated so that naive or unprimed T cells predominantly express CD45RA-positive isoforms and switch to expression of CD45RO upon activation. CD45RO expression is correlated with the memory T-cell phenotype (Akbar et al., 1988). Mice and humans lacking CD45 expression are characterized by a block of T-cell maturation (Kishihara et al., 1993; Kung et al., 2000). Jacobsen et al. (2000) investigated the role of CD45 in multiple sclerosis (MS; 126200) by measuring its expression on leukocytes. During these studies, they observed expression of a variant CD45 isoform in 1 MS patient. In contrast to other donors, all T cells and monocytes expressed the CD45RA receptor domain. The variant expression was caused by a heterozygous point mutation in exon 4 (C-to-G transversion at nucleotide 77; 151460.0001), which had been reported by Thude et al. (1995). Although the mutation did not change the encoded amino acids, it prohibited splicing of exon 4 pre-mRNA. In 3 of 4 independent case-control studies, Jacobsen et al. (2000) demonstrated an association of the mutation with MS. They found that the mutation was linked to and associated with the disease in 3 MS nuclear families. In the MS-affected members of an additional family, they found the same variant CD45 phenotype but with an unknown origin.

Timon and Beverley (2001) sequenced 2.7 kb of the 5-prime-flanking region of the CD45 gene. By CAT and EMSA analysis, they determined that the only region with promoter activity is localized within the

highly conserved first intron of the gene and is not tissue restricted. Promoter activity is strongest in the 3-prime end of intron 1, and the sequence lacks similarity with known promoters and initiators. Five-prime RACE analysis identified an alternative exon 1, designated 1a, which, like exon 1b, can be spliced to exon 2, a structure also observed in mouse.

Severe combined immunodeficiency (SCID; see 202500) is characterized by a defect in function and/or development of B and T lymphocytes, lymphopenia, and deficiency in humoral and cell-mediated immunity. Kung et al. (2000) studied a male child who presented at 2 months of age with SCID and eventually succumbed to a B-cell lymphoma at 2 years of age. Lymph node biopsies from the patient showed a lack of histologic organization and germinal center formation, and stained thin sections from the lymph node showed no expression of CD45. Indeed, CD45 expression was lacking on all leukocytes. Kung et al. (2000) identified a large deletion in one allele of the CD45 gene and a point mutation (151460.0002) in the other. A population of peripheral blood T lymphocytes was greatly diminished and unresponsive to mitogen stimulation. Despite normal B-lymphocyte numbers, serum immunoglobulin levels decreased with age.

Tchilian et al. (2001) characterized a deletion mutation in the CD45 gene of a Kurdish infant with SCID, originally reported by Cale et al. (1997), born to heterozygous, consanguineous parents. Despite successful bone marrow transplantation at age 8 months, the patient died with reactivated cytomegalovirus at age 10 months. RT-PCR and sequence analysis identified a 6-bp deletion in exon 11 of the CD45 gene (151460.0003) that resulted in the loss of glu339 and tyr340 in the first fibronectin type III module of the extracellular domain. Flow cytometric analysis demonstrated a lack of surface CD45 expression in the patient and in CHO cells transfected with the mutant cDNA but not in her parents or a healthy homozygous sib. Western blot analysis showed that deletion of the 2 amino acids results in a markedly reduced expression of the 220-kD protein. Genetic analysis of over 500 individuals from related and unrelated ethnic groups failed to detect the mutation, suggesting it is not a common polymorphism. Computational analysis of the structure of the mutant protein suggested that the lack of tyr340 destabilizes the fibronectin module, leading to unfolding and intracellular degradation. Tchilian et al. (2001) concluded that CD45 screening should be included in patients with otherwise unexplained immunodeficiency.

Majeti et al. (2000) reported the phenotype of mice with a single point mutation, glu613 to arg (E613R), that inactivated the inhibitory wedge of Cd45. The E613R mutation caused polyclonal lymphocyte activation leading to lymphoproliferation and severe autoimmune nephritis with autoantibody production, resulting in death. Both homozygotes and heterozygotes developed pathology, indicating genetic dominance of E613R. The dramatic phenotype of mice with the E613R mutation demonstrated the in vivo importance of negative regulation of CD45 by dimerization, supporting the model for regulation of CD45, and receptor-like transmembrane protein tyrosine phosphatases (RPTPs) in general.

Xu and Weiss (2002) noted that a negatively regulating ligand inducing CD45 dimerization had not been identified to that time. They hypothesized that spontaneous and isoform-differential homodimerization could offer an alternative mechanism for regulating CD45. Immunoblot analysis showed that RO isoform-enriched chemically crosslinked primary T cells or transfected cells expressing RO homodimerize more efficiently and rapidly than RAB and RABC isoforms. This homodimerization occurs in existing cell surface monomers independently of the inhibitory wedge domain and the transmembrane and intracellular domains. The dimerization efficiency of RABC increased substantially after removal of the abundant sialic acids on this isoform, but sialidase treatment did not further enhance RO homodimerization. Expression of the isoforms in O-glycosylation-defective cell lines showed that the dimerization efficiency of RABC could approach that of RO. TCR stimulation results in lower

calcium mobilization and lower soluble inositol phosphate increases in RO isoform-expressing cells compared to RABC-expressing cells. Xu and Weiss (2002) concluded that the smallest CD45 isoform, RO, homodimerizes with the highest efficiency, resulting in decreased signaling via the T cell receptor. Preferential homodimerization may account for its expression at the termination of the primary T cell response, whereas expression of RABC (RA) is required for activation of naive T cells. They proposed that these results demonstrate the biologic significance of alternative splicing and suggest a model for the regulation of receptor-like protein tyrosine phosphatase (RPTP) dimerization and function.

In a patient with familial hemophagocytic lymphohistiocytosis (603553), McCormick et al. (2003) identified a 77C-G polymorphism in exon A of the CD45 gene which caused a defect in its splicing and cosegregated with a thr435-to-met mutation in the PRF1 gene (T435M; 170280.0010). The authors postulated that both mutations were involved in the disorder.

ALLELIC VARIANTS
(selected examples)

.0001 MULTIPLE SCLEROSIS, SUSCEPTIBILITY TO [PTPRC, 77C-G]

In a patient with multiple sclerosis (MS; 126200), Jacobsen et al. (2000) identified a heterozygous C-to-G transversion at nucleotide 77 of exon 4 of the PTPRC gene. Although the mutation did not change the encoded amino acids, it prohibited splicing of exon 4 pre-mRNA. The mutation was associated with MS in 3 of 4 independent case-control studies. Jacobsen et al. (2000) found that the mutation was linked to and associated with the disease in 3 MS nuclear families. This particular variant of the PTPRC gene therefore presumably represents a susceptibility to multiple sclerosis.

Vorechovsky et al. (2001) determined allele frequencies for the 77C-G polymorphism in large numbers of MS patients and patients with common variable immunodeficiency (CVID) and IgA deficiency (IgAD) and over 1,000 controls to assess whether aberrant splicing of PTPRC caused by this polymorphism results in increased susceptibility to these diseases. They could find no difference in the frequency of the 77G allele in patients and controls in these disorders with a strong autoimmune component in etiology.

Likewise, Barcellos et al. (2001) found no evidence of genetic association between the PTPRC polymorphism and MS susceptibility or disease course in an extensive evaluation using large family-based and case-controlled comparisons.

Wood et al. (2002) found no evidence of association between this mutation and susceptibility to type 1 diabetes mellitus (222100) or Graves disease (275000). Johanneson et al. (2002) found no evidence of association between this mutation and susceptibility to systemic lupus erythematosus (152700).

.0002 SEVERE COMBINED IMMUNODEFICIENCY DUE TO PTPRC DEFICIENCY [PTPRC, IVS13DS, G-A, +1]

In a child with severe combined immunodeficiency (SCID; see 202500), Kung et al. (2000) identified compound heterozygosity at the PTPRC gene: the allele inherited from the mother carried a large deletion, while the other allele had a G-to-A transition at position +1 of the donor splice site of intron 13. Since the father did not carry the mutation, the allele had presumably undergone spontaneous mutation (although, for the privacy of the family, paternity was not proven genetically).

.0003 SEVERE COMBINED IMMUNODEFICIENCY DUE TO PTPRC DEFICIENCY [PTPRC, 6-BP DEL, NT1168]

In a Kurdish infant with SCID (see 202500), Tchilian et al. (2001) identified a 6-bp deletion at nucleotide 1168 in exon 11 of the PTPRC gene, leading to the deletion of 2 amino acids in the extracellular domain fibronectin type III module. The mutation resulted in a lack of surface PTPRC expression.

REFERENCES

1. Akao, Y.; Utsumi, K. R.; Naito, K.; Ueda, R.; Takahashi, T.; Yamada, K. :
Chromosomal assignments of genes coding for human leukocyte common antigen, T-200, and lymphocyte function-associated antigen 1, LFA-1 beta subunit. *Somat. Cell Molec. Genet.* 13: 273-278, 1987.
PubMed ID : 2955527

2. Akbar, A. N.; Terry, L.; Timms, A.; Beverley, P. C.; Janossy, G. :
Loss of CD45R and gain of UCHL1 reactivity is a feature of primed T cells. *J. Immun.* 140: 2171-2178, 1988.
PubMed ID : 2965180

3. Barcellos, L. F.; Caillier, S.; Dragone, L.; Elder, M.; Vittinghoff, E.; Bucher, P.; Lincoln, R. R.; Pericak-Vance, M.; Haines, J. L.; Weiss, A.; Hauser, S. L.; Oksenberg, J. R. :
PTPRC (CD45) is not associated with the development of multiple sclerosis in U.S. patients. *Nature Genet.* 29: 23-24, 2001.
PubMed ID : 11528386

4. Cale, C. M.; Klein, N. J.; Novelli, V.; Veys, P.; Jones, A. M.; Morgan, G. :
Severe combined immunodeficiency with abnormalities in expression of the common leucocyte antigen, CD45. *Arch. Dis. Child.* 76: 163-164, 1997.
PubMed ID : 9068311

5. Charbonneau, H.; Tonks, N. K.; Walsh, K. A.; Fischer, E. H. :
The leukocyte common antigen (CD45): a putative receptor-linked protein tyrosine phosphatase. *Proc. Nat. Acad. Sci.* 85: 7182-7186, 1988.
PubMed ID : 2845400

6. Fernandez-Luna, J. L.; Matthews, R. J.; Brownstein, B. H.; Schreiber, R. D.; Thomas, M. L. :
Characterization and expression of the human leukocyte-common antigen (CD45) gene contained in yeast artificial chromosomes. *Genomics* 10: 756-764, 1991.
PubMed ID : 1832411

7. Fischer, E. H.; Charbonneau, H.; Tonks, N. K. :
Protein tyrosine phosphatases: a diverse family of intracellular and transmembrane enzymes. *Science* 253: 401-406, 1991.
PubMed ID : 1650499

8. Goff, L. K.; van Soest, S.; Timon, M.; Tchilian, E.; Beverley, P. C. L. :
Protein tyrosine phosphatase receptor type C polypeptide (PTPRC) on human chromosome band 1q31-q32 localizes with marker D1S413 on a 610-kb yeast artificial chromosome. *Cytogenet. Cell Genet.* 87: 223-224, 1999.
PubMed ID : 10702677

9. Irie-Sasaki, J.; Sasaki, T.; Matsumoto, W.; Opavsky, A.; Cheng, M.; Welstead, G.; Griffiths, E.; Krawczyk, C.; Richardson, C. D.; Aitken, K.; Iscove, N.; Koretzky, G.; Johnson, P.; Liu, P.; Rothstein, D. M.; Penninger, J. M. :
CD45 is a JAK phosphatase and negatively regulates cytokine receptor signalling. *Nature* 409: 349-354, 2001.
PubMed ID : 11201744

10. Jacobsen, M.; Schweer, D.; Ziegler, A.; Gaber, R.; Schock, S.; Schwinzer, R.; Wonigeit, K.; Lindert, R.-B.; Kantarci, O.; Schaefer-Klein, J.; Schipper, H. I.; Oertel, W. H.; Heidenreich, F.; Weinshenker, B. G.; Sommer, N.; Hemmer, B. :
A point mutation in PTPRC is associated with the development of multiple sclerosis. *Nature Genet.* 26: 495-499, 2000.
PubMed ID : 11101853

11. Johanneson, B.; Lima, G.; von Salome, J.; Alarcon-Segovia, D.; Collaborative Group on the Genetics of SLE; BIOMED II Collaboration on the Genetics of SLE and Sjogrens Syndrome; Alarcon-Riquelme, M. E. :
A major susceptibility locus for systemic lupus erythemathosus maps to chromosome 1q31. *Am. J. Hum. Genet.* 71: 1060-1071, 2002.
PubMed ID : 12373647

12. Kishihara, K.; Penninger, J.; Wallace, V. A.; Kundig, T. M.; Kawai, K.; Wakeham, A.; Timms, E.; Pfeffer, K.; Ohashi, P. S.; Thomas, M. L. :
Normal lymphocyte development but impaired cell maturation in CD45-exon6 protein tyrosine phosphatase-deficient mice. *Cell* 74: 143-156, 1993.
PubMed ID : 8334701

13. Kung, C.; Pingel, J. T.; Heikinheimo, M.; Klemola, T.; Varkila, K.; Yoo, L. I.; Vuopala, K.; Poyhonen, M.; Uhari, M.; Rogers, M.; Speck, S. H.; Chatila, T.; Thomas, M. L. :
Mutations in the tyrosine phosphatase CD45 gene in a child with severe combined immunodeficiency disease. *Nature Med.* 6: 343-345, 2000.
PubMed ID : 10700239

14. Majeti, R.; Xu, Z.; Parslow, T. G.; Olson, J. L.; Daikh, D. I.; Killeen, N.; Weiss, A. :
An inactivating point mutation in the inhibitory wedge of CD45 causes lymphoproliferation and autoimmunity. *Cell* 103: 1059-1070, 2000.
PubMed ID : 11163182

15. McCormick, J.; Flower, D. R.; Strobel, S.; Wallace, D. L.; Beverley, P. C. L.; Tchilian, E. Z. :
Novel perforin mutation in a patient with hemophagocytic lymphohistiocytosis and CD45 abnormal splicing. *Am. J. Med. Genet.* 117A: 255-260, 2003.

16. Ralph, S. J.; Thomas, M. L.; Morton, C. C.; Trowbridge, I. S. :
Structural variants of human T200 glycoprotein (leukocyte-common antigen). *EMBO J.* 6:

1251-1257, 1987.
PubMed ID : 2956090

17. Ravetch, J. V.; Luster, A. D.; Weinshank, R.; Kochan, J.; Pavlovec, A.; Portnoy, D. A.; Hulmes, J.; Pan, T.-C. E.; Unkeless, J. C. :
Structural heterogeneity and functional domains of murine immunoglobulin G Fc receptors. *Science* 234: 718-725, 1986.
PubMed ID : 2946078

18. Saga, Y.; Tung, J.-S.; Shen, F.-W.; Boyse, E. A. :
Sequences of Ly-5 cDNA: isoform-related diversity of Ly-5 mRNA. *Proc. Nat. Acad. Sci.* 83: 6940-6944, 1986.
PubMed ID : 2944116

19. Seldin, M. F.; D'Hoostelaere, L. A.; Steinberg, A. D.; Saga, Y.; Morse, H. C., III :
Allelic variants of Ly-5 in inbred and natural populations of mice. *Immunogenetics* 26: 74-78, 1987.
PubMed ID : 2886425

20. Seldin, M. F.; Morse, H. C.; LeBoeuf, R. C.; Steinberg, A. D. :
Establishment of a molecular genetic map of distal mouse chromosome 1: further definition of a conserved linkage group syntenic with human chromosome 1q. *Genomics* 2: 48-56, 1988.
PubMed ID : 3384439

21. Shen, F.-W.; Saga, Y.; Litman, G.; Freeman, G.; Tung, J.-S.; Cantor, H.; Boyse, E. A. :
Cloning of Ly-5 cDNA. *Proc. Nat. Acad. Sci.* 82: 7360-7363, 1985.
PubMed ID : 3864163

22. Tchilian, E. Z.; Wallace, D. L.; Wells, R. S.; Flower, D. R.; Morgan, G.; Beverley, P. C. L. :
A deletion in the gene encoding the CD45 antigen in a patient with SCID. *J. Immun.* 166: 1308-1313, 2001.
PubMed ID : 11145714

23. Thomas, M. L.; Reynolds, P. J.; Chain, A.; Ben-Neriah, Y.; Trowbridge, I. S. :
B-cell variant of mouse T200 (Ly-5): evidence for alternative mRNA splicing. *Proc. Nat. Acad. Sci.* 84: 5360-5363, 1987.
PubMed ID : 2955416

24. Thude, H.; Hundrieser, J.; Wonigeit, K.; Schwinzer, R. :
A point mutation in the human CD45 gene associated with defective splicing of exon A. *Europ. J. Immun.* 25: 2101-2106, 1995.
PubMed ID : 7621884

25. Timon, M.; Beverley, P. C. L. :
Structural and functional analysis of the human CD45 gene (PTPRC) upstream region: evidence for a functional promoter within the first intron of the gene. *Immunology* 102: 180-189, 2001.
PubMed ID : 11260323

26. Tonks, N. K.; Charbonneau, H.; Diltz, C. D.; Fischer, E. H.; Walsh, K. A. :

Demonstration that the leukocyte common antigen CD45 is a protein tyrosine phosphatase. *Biochemistry* 27: 8695-8701, 1988.
PubMed ID : 2853967

27. Tonks, N. K.; Diltz, C. D.; Fischer, E. H. :
CD45, an integral membrane protein tyrosine phosphatase: characterization of enzyme activity. *J. Biol. Chem.* 265: 10674-10680, 1990.
PubMed ID : 2162357

28. Trowbridge, I. S. :
CD45: a prototype for transmembrane protein tyrosine phosphatases. *J. Biol. Chem.* 266: 23517-23520, 1991.
PubMed ID : 1836211

29. Vorechovsky, I.; Kralovicova, J.; Tchilian, E.; Masterman, T.; Zhang, Z.; Ferry, B.; Misbah, S.; Chapel, H.; Webster, D.; Hellgren, D.; Anvret, M.; Hillert, J.; Hammarstrom, L.; Beverley, P. C. :
Does 77C-G in PTPRC modify autoimmune disorders linked to the major histocompatibility locus? *Nature Genet.* 29: 22-23, 2001.
PubMed ID : 11548742

30. Wood, J. P.; Bieda, K.; Segni, M.; Herwig, J.; Krause, M.; Usadel, K. H.; Badenhoop, K. :
CD45 exon 4 point mutation does not confer susceptibility to type 1 diabetes mellitus or Graves' disease. *Europ. J. Immunogenet.* 29: 73-74, 2002.

31. Xu, Z.; Weiss, A. :
Negative regulation of CD45 by differential homodimerization of the alternatively spliced isoforms. *Nature Immun.* 3: 764-771, 2002.
PubMed ID : 12134145

CONTRIBUTORS

Victor A. McKusick - updated : 4/7/2003
Victor A. McKusick - updated : 12/23/2002
Paul J. Converse - updated : 7/23/2002
Victor A. McKusick - updated : 8/24/2001
Paul J. Converse - updated : 5/14/2001
Carol A. Bocchini - updated : 3/6/2001
Paul J. Converse - updated : 2/16/2001
Paul J. Converse - updated : 2/16/2001
Ada Hamosh - updated : 1/19/2001
Stylianos E. Antonarakis - updated : 1/3/2001
Victor A. McKusick - updated : 11/27/2000

CREATION DATE

Victor A. McKusick : 6/15/1987

EDIT HISTORY

alopez : 6/10/2003
tkritzer : 4/7/2003
tkritzer : 12/26/2002
terry : 12/23/2002
alopez : 8/6/2002
alopez : 7/23/2002
alopez : 4/24/2002
terry : 8/24/2001
mgross : 5/14/2001
terry : 3/6/2001
carol : 3/6/2001
mgross : 2/21/2001
mgross : 2/21/2001
terry : 2/16/2001
terry : 2/16/2001
mcapotos : 1/30/2001
carol : 1/22/2001
terry : 1/19/2001
mgross : 1/3/2001
mgross : 11/29/2000
mgross : 11/28/2000
mgross : 11/28/2000
terry : 11/27/2000
dkim : 7/23/1998
dkim : 7/23/1998
psherman : 4/10/1998
mark : 12/4/1995
carol : 3/12/1994
supermim : 3/16/1992
carol : 2/21/1992
carol : 2/12/1992
carol : 6/6/1991
supermim : 6/4/1991

ALLELIC VARIANTS
(selected examples)

- 0001 : MULTIPLE SCLEROSIS, SUSCEPTIBILITY TO
  - Mutation : PTPRC, 77C-G
- 0002 : SEVERE COMBINED IMMUNODEFICIENCY DUE TO PTPRC DEFICIENCY
  - Mutation : PTPRC, IVS13DS, G-A, +1
- 0003 : SEVERE COMBINED IMMUNODEFICIENCY DUE TO PTPRC DEFICIENCY
  - Mutation : PTPRC, 6-BP DEL, NT1168